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CLINICAL INFORMATION

Anaesthesia in a child operated for cleft lip associated with Patau's syndrome

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Abstract Patients with Patau's syndrome (Trisomy 13) have multiple craniofacial, cardiac, neurological and renal anomalies with very less life expectancy. Among craniofacial anomalies cleft lip and palate are common. These craniofacial and cardiac anomalies present difficulties with anaesthesia. We therefore describe the anaesthetic management in the case of a Trisomy 13 child for operated for cleft lip at 10 months of age.

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PALAVRAS-CHAVE

Síndrome de Patau;
Lábio leporino;
Fenda palatina;
Anestesia pediátrica

Anestesia em criança operada para lábio leporino associado à síndrome de Patau

Resumo Os pacientes com síndrome de Patau (trissomia 13) apresentam várias anomalias craniofaciais, cardíacas, neurológicas e renais, com expectativa de vida bem menor. Entre as anomalias craniofaciais, o lábio leporino e a fenda palatina são comuns. Essas anomalias craniofaciais e cardíacas apresentam dificuldades na anestesia. Portanto, descrevemos o manejo anestésico em uma criança de 10 meses de idade com Trissomia 13 submetida à cirurgia de lábio leporino.

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Introduction

Patau's syndrome is autosomal Trisomy 13, first described by Patau et al. in 1960.¹ It is a rare disorder, with incidence of 1 in 6000 live births, with multiple craniofacial, cardiac, neurological and renal anomalies.² Median survival is less than 3 months. Seventy-five percent of cases of Patau's syndrome are due to primary non-disjunction of the 13th chromosome during the first meiotic division (free trisomy; 47, XX or XY, +13); the remaining 25% is due to an unbalanced Robertson translocation involving the long arms of one 13 and one 14 chromosome e.g. 46, XX or XY, +t(13q14q).

Changes in the number or structure of chromosomes are a major cause of congenital anomalies and intellectual impairment. Trisomy 13 is the third most common autosomal Trisomy at birth, with Trisomy 21, followed by Trisomy 18, occurring more frequently.^{3,4} Intrauterine growth restriction, along with facial, heart, and limb anomalies, are the most striking features. The most common cranial abnormality diagnosed with cranial imaging in infants with Trisomy 13 is holoprosencephaly.⁴ Severe mental retardation is a characteristic feature in infants who survive.

Usual clinical presentation of patients with Patau syndrome^{5,6}

Many fetuses never survive until term and are stillborn or spontaneously abort. Features include:

Intrauterine growth restriction and low birth weight.

Congenital heart defects: these occur in 80%; they include atrial septal defect, ventricular septal defect, patent ductus arteriosus, dextrocardia.

Holoprosencephaly: the brain doesn't divide into two halves; this can present with midline facial defects including:

Cleft lip and palate.

Microphthalmia or anophthalmia.

Nasal malformation.

Hypotelorism (reduced distance between the eyes) or cyclops.

Other brain and central nervous system abnormalities including:

Neural tube defects.

Other anatomical defects of the brain.

Severe learning disability.

Problems with control of breathing (central apnoea).

Other craniofacial abnormalities include:

Microcephaly.

Scalp defects (cutis aplasia: skin missing from the scalp).

Ear malformations and deafness.

Capillary haemangiomas.

Gastrointestinal abnormalities: omphalocele, exomphalos, hernias.

Urogenital malformations: polycystic kidneys, micropenis or hypertrophy of the clitoris.

Abnormalities of hands and feet: polydactyly (extra fingers or toes), rocker-bottom feet.

Therefore, children with Trisomy 13 may not have surgeries for non-life threatening malformations, such as cleft lip and palate. Here we report a case of lip repair surgery performed for unilateral complete cleft lip and palate in a patient diagnosed with Trisomy 13 and holoprosencephaly.

Case history

The patient named Maahi aged 10 months, weighing 8 kg reported to our department for cleft lip repair. The patient was found to have blepharophimosis, telecanthus, microtia and microphthalmia on the right side with no movement at cervical spine. The examination of the face in general was found to have deficient zygomatic arch, skeletal cleft (tessier no. -13, 14). The patient had unilateral complete cleft lip and palate, hypoplasia of the nose, aplasia of the columella, a short prolabium. An ECG showed sinus rhythm, with p pulmonale and right ventricular strain/hypertrophy. The routine blood investigations were normal. The chest X ray, electrocardiogram and ultrasonography of the chest suggested dextrocardia. The ultrasonography of abdomen was not revealed any significant finding. No congenital heart diseases were detected on echocardiogram except dextrocardia. The MRI brain shows no obvious abnormalities. The CT scan of neck suggested butterfly C5-C6 vertebrae and posterior spina bifida at C3-C6 levels. The karyotyping report of this patient suggest Patau syndrome.

The patient had never undergone any major surgeries in the past and hence there were no medical records pertaining to the anaesthesia available. Due to physical disabilities and restricted neck movement predicting the difficult intubation in preoperative checkup. After attaching the standard ASA monitors ECG, NIBP, and SPO₂, general anaesthesia was planned for the correction of the cleft lip. On day of surgery, child was fasted for 4 h and atropine 0.02 mg/kg was administered intramuscular and promethazine 7.5 mL syrup given as premedication 1 h prior to the induction of anaesthesia in preoperative area. Anaesthesia was induced by sevoflurane in 100% oxygen. While maintaining spontaneous respiration, an initial assessment of the airway by direct laryngoscopy revealed a high arched cleft of hard palate with only the lobes of cleft soft palate visible without visualization of epiglottis and glottis aperture (Cormack and Lehane view grade 4), endotracheal intubation tried but because of no movement at cervical spine we were not able to intubate the child. As ventilation was possible the child was given injection succinylcholine 15 mg but still intubation was not possible. We had backup of paediatric fiberoptic bronchoscope and ENT surgeon for tracheostomy. The paediatric fiberoptic bronchoscope available was not appropriate for the patient. Thus we inserted an urological guide wire through working channel of FOB into the trachea and cuffed ETT of 4.0 mm railroad over guide wire. After confirmation of ETT with auscultation, chest rise and EtCO₂, anaesthesia was maintained with sevoflurane on spontaneous ventilation and increments of fentanyl 2 µg/kg and local infiltration of lignocaine 1% with

adrenaline (1:200 000) at surgical site. The fluid was administered according to standard 4, 2, 1 formula. On completion of surgery patient was extubated after achieving adequate level of consciousness and shift to post anaesthesia care unit.

Discussion

Trisomy 13 (Patau syndrome) is a congenital disorder first described by Patau in 1960, and it is caused by having an extra 13th chromosome.¹ Children with Patau syndrome or Trisomy 13 usually have severe mental retardation due failure of development of separate cerebral hemispheres and lateral ventricles (holoprosencephaly). The frequency of this syndrome has been reported between 1:3000 to 1:29 000 live births,^{7,8} and the incidence and survival rate in females is higher than those in males at all ages.^{9,10}

Cardiac anomalies occur in more than 80% of cases¹¹⁻¹³ and are usually multiple. Dextroposition of the aorta has been reported in 50% of cases, resulting in double outlet right ventricle and tetralogy of Fallot.¹⁴ The most frequently reported lesions are atrial and ventricular septal defects, patent ductus arteriosus, dextrocardia.¹¹

The presence of a short neck and small mouth with a high arched cleft palate may make tracheal intubation, or insertion of a laryngeal mask, difficult. Keeping in mind these potential difficulties, Aids to intubation, including a fibre-optic bronchoscope, should be available for these cases. Apnoeic episodes are also a reported feature of Patau's syndrome in 50% of cases,¹² hence opioid analgesics must be used with caution. We therefore recommend, especially in major surgical cases, that facilities for elective postoperative ventilation should be available.

The combination of the requirement for cardiovascular stability with the management of a difficult airway often makes the choice of anaesthesia technique difficult. Cardiovascular function is also further compromised by both intravenous and inhalational anaesthetic agents and they should be used cautiously to avoid precipitate changes in systemic or pulmonary vascular resistance.

There have been few reports about cleft lip and palate surgery for Trisomy 13 cases,¹⁵ despite there being many reports about cardiac procedures.^{9,16} Baty et al. showed that the percentage of patients that had surgery for Trisomy 13 during the neonatal period was 23%.⁹ Nelson et al. described that cleft palate surgery represented 3.2% ($n = 1075$) of the total major therapeutic procedures in children with Trisomy 13 that were performed from 1997 to 2009.¹⁶ The prognosis for the case that we operated for the repair of cleft lip was expected to be better than in other patients with Trisomy 13 that who had similar problems like microcephaly, facial clefts, short neck, holoprosencephaly etc. The features of her cleft lip such as hypoplasia of the prolabium or aplasia of the columella (as mentioned earlier) made the lip repair surgery more difficult. However, we finished the operation in the appropriate amount of time (approximately 45 min) by careful preoperative planning. The patient has made steady progress since the operation.

Conclusion

Despite the complications related to Trisomy 13 it possible to conduct short, low-invasive procedures which are carefully preplanned. Surgical corrections and other surgeries in patients with Trisomy 13 might tend to pose difficulties as far general anaesthesia is concerned but these complications can be managed if there is careful pre-evaluation of the patient.

Conflicts of interest

The authors declare no conflicts of interest.

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